CASE REPORT

A Case of Limb Body Wall Complex – A Rare Foetal Malformation

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ABSTRACT

We report a case of very rare congenital anomaly "limb-body wall complex" type-II (ventral wall defect) also known as "body stalk syndrome" in a 15 weeks foetus. On antenatal 3-D ultrasound scan there was parieto-occipital encephalocele, complete absence of the right upper limb, gastroschisis and scoliosis seen at the thoraco-lumbar spine.

There is no cure for limb-body wall complex, and it is also incompatible with life. Severe pulmonary hypertension and respiratory failure are the most common cause of death in case of LBWC. Survival of newborn with LBWC is rare. In our case pregnancy was terminated at 15 weeks and all antenatal USG scan findings were confirmed after delivery of foetus.

KEY WORDS: Body stalk syndrome, Encephalocele, Gastroschisis, Omphalocele, Limb-body wall complex, Scoliosis.

INTRODUCTION

Limb body wall complex (LBWC) is an entity characterized by the presence of an abdominal wall defect associated with variable spectrum of limb and visceral anomalies[1]. The diagnosis of this entity is based on two of the three following characteristics:

(1) Exencephaly/ Encephalocele and facial clefts;

(2) Thoraco - and/or Abdominoschisis; and

(3) Limb defects.

The diagnosis of limb– body wall complex can usually be made on prenatal USG by the end of the first trimester[2].

Limb–body wall complex is primarily differentiated from other entities associated with abdominal wall defects based on the position of the defect with respect to the umbilical cord insertion. The ventral wall defect in limb–body wall complex commonly consists of a large, eccentric, lateral abdominal wall defect with the abdominal organs directly attached to the placenta or the uterine wall[3].

Two adhesion phenotypes have been described, the "placentocranial" and "placentoabdominal." LBWC is also known as "body stalk syndrome." Unfortunately, there is no cure for LBWC, and it is generally considered to be incompatible with life (fatal)[4].

CASE REPORT

A 32 year old primigravida with an uneventful antenatal period and 4 month amenorrhoea was referred to radiodiagnosis department for routine antenatal USG. There was no history of consanguinity or any family history of malformation. There was no history of ingestion of nonprescribed drugs or use of alcohol or tobacco in any form throughout the whole duration of the pregnancy. No past history of COVID-19 positive. She had normal blood profile and O +ve blood group. Haemoglobin was 9.3 gm/dl, TLC was 12000 per cubic mm and AFP level was increased and it was 35.5 MoM.

The 3-D ultrasonography revealed femur length corresponding to 15 weeks 3 days and other associated multiple foetal anomalies.

There was a large anterior abdominal wall defect through which abdominal content were herniating into the amniotic fluid (gastroschisis). The eviscerated organs form a complex and bizarre appearing mass, which consist of the liver, the stomach and the bowel loops shown in the figure 1.

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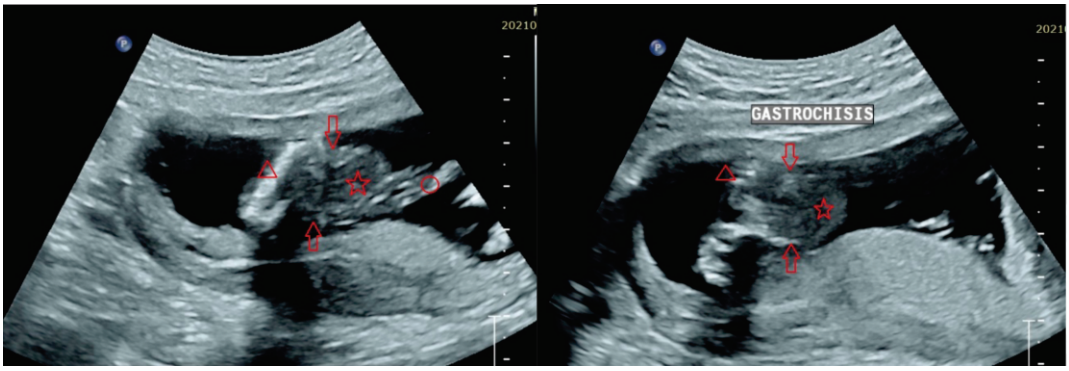

Figure 1: Showing gastrischisis. (Arrow – abdominal wall defect, star – bizarre mass consist of the silver, the stomach and the bowel loops, triangle- spine, circle- umbilical cord)

There was 9 mm defect noted at the parieto-occipital region from which brain tissue was herniating into the membrane covered sac consistent with encephalocele shown in figure 2.

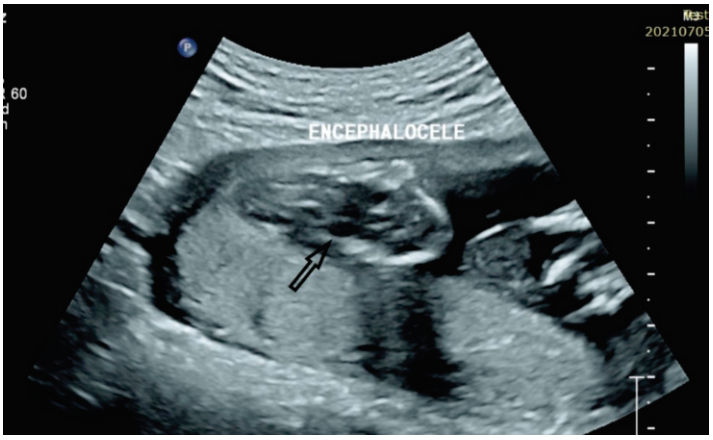

Figure 2: Showing defect (arrow) in parieto-occipital region through which meanings with brain tissue are herniating – consistent with encephalocele.

Further study revealed scoliosis at the thoraco-lumbar spine shown in figure 3, but there was no evidence of spinal meningocele.

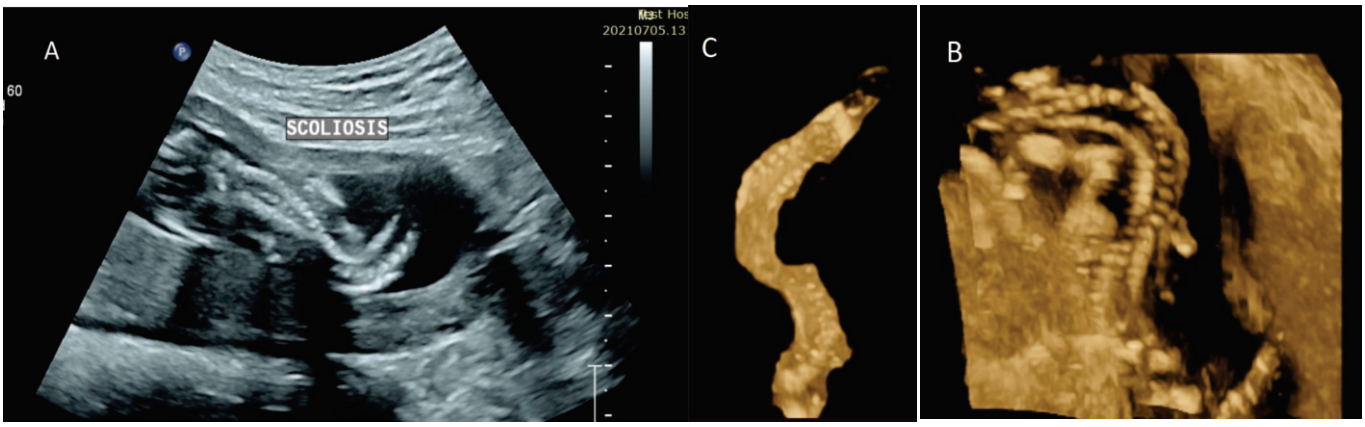

Figure 3: Showing scoliosis of thoracolumbar spine (A-2D image, B&C images)

There was complete absence of the right upper limb shown in figure 4. Left upper and both lower limbs appear normal.

Figure 4 : Showing that right humerus is absent and left humerus is present.

3 vessels umbilical cord was short and malformed. It was embedded into amniotic sheet, which was connected between placenta and skin margin of anterior body wall defect.

The patient and her family were informed about the poor prognosis of foetus due to this anomaly and they decided to terminate this pregnancy. Therefore she was referred back to department of Gynecology and Obstetrics for further management with the diagnosis of LBWC.

Medical termination of pregnancy was done by Gynecology and Obstetrics department under all aseptic precautions by induced expulsion of the foetus weighted approx. 100 grams. All the antenatal findings were confirmed after delivery of foetus as shown in figure 5.

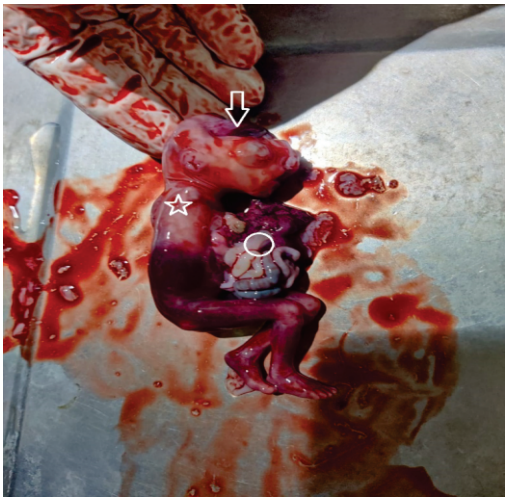

Figure 5 : Showing delivered baby with encephalocele (arrow), gastroschisis (circle), absent right upper limb (star).

DISCUSSION

LBWC also known as Body stalk anomaly, is a sporadic, lethal abnormality with a reported incidence of 1 in 14,000–42,000 pregnancies[5]. The incidence at birth is, however, lesser and it is about 0.2–0.3/100,000 live births because the majority of the affected foetuses undergo intrauterine deaths[6].

The diagnosis of this entity is based on two of the three following characteristics: (a) exencephaly/ encephalocele and facial clefts; (b) thoraco - and/or abdominoschisis; and (c) limb defects. A definite association with internal anomalies and severe kyphoscoliosis makes a more distinct concept of the pathogenesis reasonable[3]. In our case, abdominoschisis was present with limb defects and scoliosis. Oligohydramnios may be present during the second and third trimesters, in which case magnetic resonant imaging may be helpful to elucidate the anatomic structure[7].

The exact etiology of this condition is still unclear, Tropin's amniotic band theory, and Van Allen's vascular theory failed to explain all the anomalies in LBWC[3]. The pathogenesis is uncertain, but three pathogenetic mechanisms have been proposed which are: abnormal folding of the trilaminal embryo during the first 4 weeks of development, early amnion rupture with amniotic band syndrome, and early generalized compromise of embryonic blood flow[2]. The anomaly can be classified into two based on the malformation phenotypes. Type-I is based on craniofacial defect whereas Type-II is recognized based on the ventral wall defects[2,6]. This case was diagnosed as type-II, ventral wall defect.

There is no correlation of LBWC with the foetal gender, parents' age, or karyotype anomalies[3].

The diagnosis of this condition can be established by measuring maternal serum alpha fetoprotein levels. Prenatal ultrasound examination can detect this anomaly as early as first trimester (usually by the end of first trimester)[8].

In view of the fact that LBWC is incompatible with life, it is important to diagnose the condition early in pregnancy to help the parents make an informed decision. Prenatal ultrasound scan in the second trimester does not only confirm the diagnosis but also differentiates it from the other nonlethal dysplasias such as gastroschisis, omphalocele, cloacal dystrophy, and urachal cyst as well as from other lethal polymalformation complexes. Such complexes include pentalogy of Cantrell, omphalocele-exstrophy-imperforate anus-spinal defects, amniotic band sequence, and omphalocele with cloacal–bladder exstrophy complex[3,5].

Most pregnancies with LBWC lead to miscarriages, termination of pregnancies, or still births7,9. Death usually occurs due to pulmonary hypoplasia which leads to respiratory failure and severe pulmonary hypertension. Survival of newborn with this condition is quite rare, although there have been cases whereby the baby survived up to 84 days[7]. The pregnancy in this index case was terminated at 15 weeks.

Learning Points

a) LBWC is a very rare fatal anomaly and few cases are reported in literature.
b) Whenever we observe abdominoschisis, limb defect and scoliosis on ultrasonography, suspicion of LBWC should be raised.
c) Differentiation with other abdominal wall defects such as gastrochisis and omphalocele is necessary to make a diagnosis of LBWC.
d) Early diagnosis of LBWC should be done to prevent maternal health.
e) Once LBWC is diagnosed antenatally, pregnancy should be terminated.

CONCLUSION

The prognosis of LBWC is very poor as compared to isolated gastroschisis and exomphalos. To reduce the maternal morbidity and mental trauma to family, pregnancy should be terminated after establishing correct diagnosis, which requires careful ultrasound of the foetus whenever ventral abdominal wall defect is suspected.

The patient was informed of the poor prognosis and after counselling she accepted the termination of the pregnancy.

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